Epilepsy-telangiectasia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Epilepsy-telangiectasia syndrome. ORPHA:1951

Epilepsy telangiectasia syndrome is characterized by intellectual deficit, epilepsy, palpebral conjunctival telangiectasias and diminished serum IgA, particular facies and a shortened fifth finger. It has been reported in six siblings from a Mexican family. It is probably transmitted as an autosomal recessive trait.